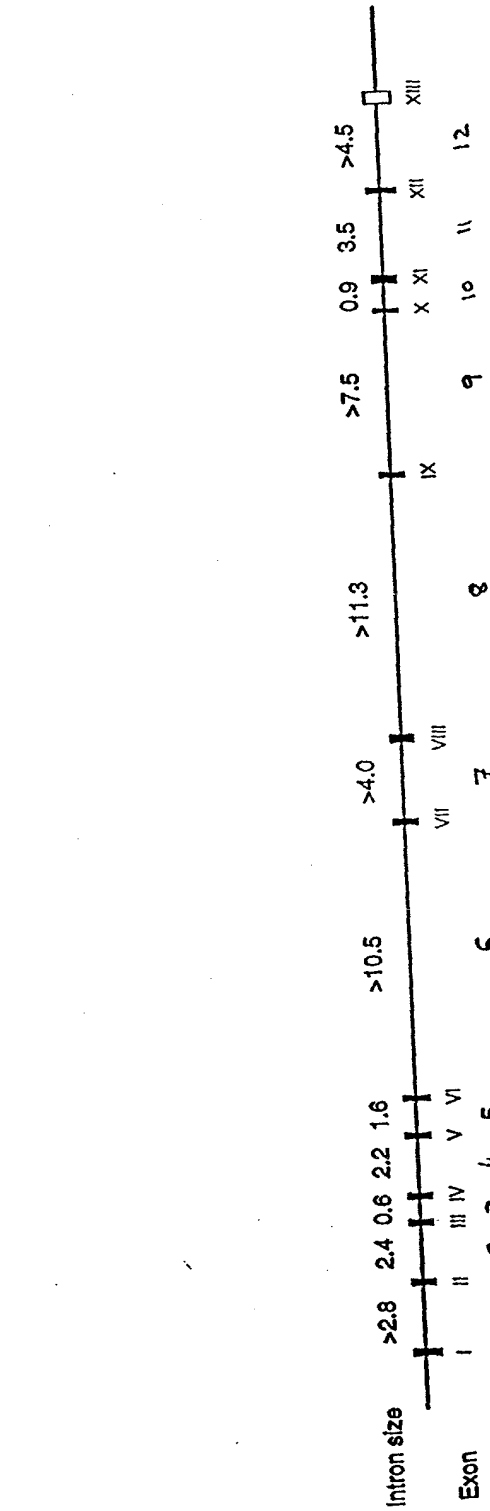
Figure 1


Intron size
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2.4
0.6
2.2
1.6
>10.5
>4.0
>11.3
>7.5
0.9
3.5
>4.5
Exon
Intron
I
II
III
IV
V
VI
VII
VIII
IX
X
XI
XII
XIII
1
2
3
4
5
6
7
8
9
10
11
12
5 kb

Figure 2A

promoter and exon 1

ACTGCGGAGATGAGGGTCTAGAAGGTGGTGGCGGGGCAT
GTGGACCGTTGTAAGGGCTCTGGGG**TTCCTGGGTGGCT**
GGCGAAGTCCTACTCACAGTGACCAACCATGATGATGGT
CCCGATAGAGGAGGAGAGGGAGGAGGAGGGAAAAGGAAG
GGTGAGGGGCTCAGAGGGGAGAGCTGGGAGGAGGGGAGA
CATAGGTGGGGGAAGGGGTAGGAGAAAGGGGAAGGGAGC
AAGAGGGTGAGGGGCACCAGGCCCCATAGACGTTTTGGC
TCAGCGGCCACGAGG**CTTCATCAGCTCCCGCCCCAAAAC**
GGAAGCGAGGCCGTGGGGGCAGCGGCAGCATGGCGGGGC
TTGTCTTGGCGGCCATGGCCCCGCCCCCTGCCCGTCCGA
TCAGCGCCCCGCCCCGTCCCCGCCCCGACCCCGCCCCGG
GCCCGCTCAGGCCCCGCC**CCTGCCGCCGGAATCCTGAAG**
CCCAAGGCTGCCCGGGGGCGGTCCGGCGGCGCCGGCGAT
GGGGC*ATAAAA*CCACTGGCCACCTGCCGGGCTGCTCC

*TGCGTGCGCTGCCGTCCCGGATCC*ACCGTGCCTCTGCGG
CCTGCGTGCCCGGAGTCCCCGCCTGTGTCGTCTCTGTCG
CCGTCCCCGTCTCCTGCCAGGCGCGGAGCCCTGCGAGCC
GCGGGTGGGCCCCAGGCGCGCA**GACATGGCTGCTCCGC** A
CAAAGCGCGCTGGGCTGCCGGGGCGCTGGGCGTCGCGGG
GCTACTGTGCGCTGTGCTGGGCGCTGTCATGATCGTGAT
GGTGCCGTCGCTCATCAAGCAGCAGGTCCTTAAG

GTGGGTGAGGGAGACCCCAGGGGGTCCGCGCACGGACCC
GGGCTGTTGGGCGCTGGGCGCCGGGAGGACCCGCGCGTT
GCGGTGGGTGGGCGACCGCAGCGGAATCGGCGCCCGGGC
CTGGCGCCGCAGAACACGAGGGAGGCCAGGCGCTTCGGG
AGGGGCTGCTGCCCGCCTCCCCACCACCCTCACC

Figure 2B

exon 2

AGCCTCATGTGCGAAGGGCTTTCCCACCACCTCCTATCC
CAAGCTCCCGCCGAGGAGCCCCTTCCCTGGCCGGGCTCG
GGCAGCTGTTCCGGAGCCTTGTGGTGGGGCGTGGGGCC
CTCATCACTCTCCTCACAAGCGTACTTGTCCCTTCCC
CTGCAG

AACGTGCGCATCGACCCCAGTAGCCTGTCCTTCAACATG
TGGAAGGAGATCCCTATCCCCTTCTATCTCTCCGTCTAC
TTCTTTGACGTCATGAACCCCAGCGAGATCCTGAAGGGC
GAGAAGCCGCAGGTGCGGGAGCGCGGGCCCTACGTGTAC
AG

GTGAGGCTGTGTCCACGTGATGGTGGACGGGCCGGCTGA
CGCTGGGCATGGGACGGGTCTCA**NAGTGGACGGGATG**
GGGAGGCTGCTGACTGACCCCCAAACATTGTTCCGGAA
GCACGCAACTCATAGTCGGGGTAAGTGCTACTCCCAAAA
AAGTTTGCGT

exon 3

CATGTCCTGCAGTGGGCAGGCAGCGGGAGGGACAGACTT
GGCGAAGGGGCCGAGCTCAGCTTTGGCTGTGGGGCCGGA
GGTGTGCACAGACGTCCAGGGCCCCTGGTTCCCAGGCAG
GC**ATTGCAGGCGAGTAGAAG**GGAAACGTCCCATGCAG
CGGGGCGGGGCGTCTGACCCACTGGCTTCCCCCACAG

GGAGTTCAGGCACAAAAGCAACATCACCTTCAACAACAA
CGACACCGTGTCCTTCCTcGAGTACCGCACCTTCCAGTT
CCAGCCCTCCAAGTCCCACGGCTCGGAGAGCGACTACAT
CGTCATGCCCAACATCCTGGTCTTG
A
GTGAGGCTGCCCTGTGGCCCACGCCGCCTCGCACCCTGA
CCTCGTCCCC**TGTCTCTCCTCCCGCCTG**CCCCTTGTG
CAGAGAGCAGTCCCTGAGGTGGTCGGAGCGTGGGGACTC
ACGCCTGGTGGGTGGCTTTCGGCCCTGTGCTGTCTCCAC
CACCCCCA

Figure 2C

exon 4

GGTGGTTCTGGTGTCCCAGATGCCCCACGTGGCCACTCC
AGGGGCCTCCTGCACCCCAGCATTTCCCTTCATGGGCT
CTTTGCTGTGAGGCCCAGCTGGGGCCAAGGGAGGATG
GGCCAGCCACGTCCAGCCTCTGACACTAGTGTCCCTTCG
CCTTGCAG

GGTGCGGCGGTGATGATGGAGAATAAGCCCATGACCCTG
AAGCTCATCATGACCTTGGCATTCACCACCCTCGGCGAA
CGTGCCTTCATGAACCGCACTGTGGGTGAGATCATGTGG
GGCTACAAGGACCCCTTGTGAATCTCATCAACAAGTACT
TTCCAGGCATGTTCCCCTTCAAGGACAAGTTCGGATTAT
TTGCTGAG

GTACGTGTGGCCTGGTGAGAAGCCAAAGATTCAGGCCTG
TGTCCTGT**CTTCCCCTCACACAGCCTGG**ACACTGGTC
ACCAGCTTGCTTTGTAGCTGGCTGGGGATCTAGTGGCTG
TGGGTTGTAAGTGACTGAGAACCTGACTCAAACCGGCTT
GAGTGAAA

exon 5

CCTCTCGGTCCCCAGACACTGGGCATTTGGCAGTGAACC
AGATGCTGGGGGCCCTGTCCTTCTGGTGGAGGGGGAGGA
GGGCTCA**GCCCAGAATGTTCAGACCAG**GCCGGCTCAA
TGGCAGGCCTAAGCCTTACGATGCTGTTCCCTGCTGTGT
CTGTAG

CTCAACAACTCCGACTCTGGGCTCTTCACGGTGTTCACG
GGGGTCCAGAACATCAGCAGGATCCACCTCGTGGACAAG
TGGAACGGGCTGAGCAAG

GTGAGGGGCGAGAGGCGAGGGCCCCTGTCGCCAGGGAGA
GGGGAGGGTGGGCC[]GGCCATGGCTGCTCGGGAGTGGCA
GGGACCAGAGAGCTCCTTCTTC**CTTTGTCGTGAAGAG**
GGTGCTGGGAGGATGAACACTCTTGAAGTTGGAGGAGGG
ATTTTA

T

Figure 2D

exon 6

TCTCTGTGTGTCTACATAGCCTGCCCTCTTCCCACCGTG
CCAGTATTGGGAATTGAGTGGCCGTGCGTGCACCAGGGT
GAGTTAGGTGTGCAG**CACCTGAGAGGGCTTATTA**AGG
GGCCTTGGCCCTACTGAGGGGTCTAGTCTGGATGCTTCC
CCCCAG

GTTGACTTCTGGCATTCCGATCAGTGCAACATGATCAAT
GGAACTTCTGGGCAAATGTGGCCGCCCTTCATGACTCCT
GAGTCCTCGCTGGAGTTCTACAGCCCGGAGGCCTGCCG

GTAATCACTGGGACTCGGGGCCTCCTGGGTTTCCTGGGT
AGCTCATGGCCAAATTCTGTGGTGTTGGCTGT**GCACTT**
GGAAAGCATTTTGACTCATCGTGGATTTGACTCAGTAG
CCCTTGGCACCAGCTTGAATTCTCTTTGGTCACACCACC
AAAAGC

exon 7

GGAGGTCGCTGCAGCTCCGCGGGTGAGAGATGGGGGCGG
TTTGGACCCGGGAGGTGGTAGCGCCCGTGGGGAGAAGTG
GCTGGATCTGGGCAGCCTTTGGCAGGGCCTGGCTCTGGC
CGCCGGGTCTGGGTGTCCCCTCTCATCCTGTCTGTCC
CCTGCAG

ATCCATGAAGCTAATGTACAAGGAGTCAGGGGTGTTTGA
AGGCATCCCCACCTATCGCTTCGTGGCTCCCAAAACCCT
GTTTGCCAACGGGTCCATCTACCCACCCAACGAAGGCTT
CTGCCCGTGCCTGGAGTCTGGAATTCAGAACGTCAGCAC
CTGCAGGTTCA

GTACGTGCCGTCCCCTGTTCTGGGATNGCCGGAGGGTGT
TAGGTNTNGGGCACCTNANGGTTTATCTGCCCAATGCTG
TCTGCTTAATCTCTGGCCTCTGTACTCTTGATAACC
CATTAAGCCAAAAATATGATGCCTCTGGGACGATATCTG

Figure 2E

exon 8

TGGGGCTTTTTACAGAATGGAGGAAGGGATCCTCTCT
GTCGGGTATTATGGTCATCGCCACGGGGGTGCCGTGCAG
ACCACAGCTCTGTGCAGACTTCCGGAGTGGCAGGACGTG
CCAATATACTGTCGTTGTATGATGTCCCCTCCCTGCCCT
TGTTGTAG

GTGCCCCCTTGTTTCTCTCCCATCCTCACTTCCTCAACG
CCGACCCGGTTCTGGCAGAAGCGGTGACTGGCCTGCACC
CTAACCAGGAGGCACACTCCTTGTTCCTGGACATCCACC
CG

GTGAGCCCCTGCCATCCTCTGTGGGGGGTGGGTGATTCC
TGGTTGGAGCACACCTGGCTGCCTCCTCTCTCCCCAG
GCAGAGAGCTGCTGTGGGCTGGGGTGGTGGGAAGCCTGG
CTTCTAGAATCTCGAGCCACCAAAGTTCCTTACT

exon 9

CCCCAGCCTGTGGCTTGTTTTAGGTAAGATACAAGCAAG
CTCCACTGGGCAGTTAGCTGGGACGCCCACCCTCTTGAC
TGGGACCAGGGAAAAGAAGGTTGACTGTGTCCCTGGA
GCTTGGGGGTGGCCAGTCTCCTCACTGTGTTTGTTGCCG
CAG

GTCACGGGAATCCCCATGAACTGCTCTGTGAAACTGCAG
CTGAGCCTCTACATGAAATCTGTCGCAGGCATTGG

GTGAGTGGGGACTGGGAACTGGGGCTGCATTGCTCATTG
AGAGATTANGTGCTCAGTGCTCCAGTGTTCCCAGAC
TCCCCTGACATACCCCAGGAAACAGGGCATGGGGAAGGG
AGAGGGTCCTATTGGGGGTGGAATCCAGTCCCTGCTGAT
CTTCTC

Figure 2F

exon 10

ATGGCTCCTAAAGTGTTTCAGCTCATTGTTTATATTTGG
TGGTGAGGGTTTAGTGTGTGCAAAATTATACTAAACC
TGTTTAGATGTTGTATTCAAGCAGAATTAGATCAAGTTT
GGGTGTAAGACTTTGTTCCAACACCTATGTCTTGCTTAT
TTCCAG

ACAAACTGGGAAGATTGAGCCTGTGGTCCTGCCGCTGCT
CTGGTTTGCAGAG

GTAAGGGTGCGTTGGGCACAGCGTCGGGGGCTTTTGTTA
ATAGCCAATGTGGGCATTTGAGGCAGGAGGCGGGGGG
AGCACCTTGTAGAAAGGGAGAGGGCTGAGCCAGGGTAAC
CGGACTGTTACATGGACCAGCGTATCATACACTTCACCC
TGTC

exon 11

CCTGGAGGGAGGAGGTCCCTGGCAGGCTCCAACACATGC
TTTAGCCGGGAAGCTTGAGGTGGGGAAAAGCTGAGGCGG
GCACAGAGGAAGGTGTTGGGTGGCATCTGCGCTGTAG
CCCGCAGCCTGCGGCCCCAGCTCATGTGTTTGTCATTCT G
GTCTCCTCAG

AGCGGGGCCATGGAGGGGGAGACTCTTCACACATTCTAC
ACTCAGCTGGTGTTGATGCCCAAGGTGATGCACTATGCC
CAGTACGTCCTCCTGGCGCTGGGCTGCGTCCTGCTGCTG
GTCCCTGTCATCTGCCAAATCCGGAGCCAA

GTAGGTGCTGGCCAGAGGGCAGCCCGGGCTGACAGCCAT
TCGCTTGCCTGCTGGGGGAAAGGGGCCTCAGATCGGACC
CTCTGGCCAACCGCAGCCTGGAGCCCACCTCCAGCAG
CAGTCCTGCGTCTCTGCCGGAGTGGGAGCGGTCACTGCT
GGGGG

Figure 2G

exon 12

CCCCACATCTCAGCCACCTGCAATCGTTGAGGGTTGTTG
GACTCTAAACTTATGTGCCTTTCCTGTTTCCTCTTTGCC
TTTTGCAAATTGAAGAACCGTGTAAAACCATTTTTAT
GTGGCTTCAACGTCAACTATAAATTAGCTTGGTTATCTT
CTAG

GAGAAATGCTATTTATTTTGGAGTAGTAGTAAAAAGGGC
TCAAAGGATAAGGAGGCCATTCAGGCCTATTCTGAATCC
CTGATGACATCAGCTCCCAAGGGCTCTGTGCTGCAGGAA
GCAAAACTGTAG

GTGGGTACCAGGTAATGCCGTGCGCCTCCCCGCCCCCTC
CCATATCAAGTAGAATGCTGGCGGCTTAAAACATTTGGG
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TGTCTACAGACTGAAGATGCATATTTGTGTATTTTGCTT
TTGGAGAAA

Figure 3A

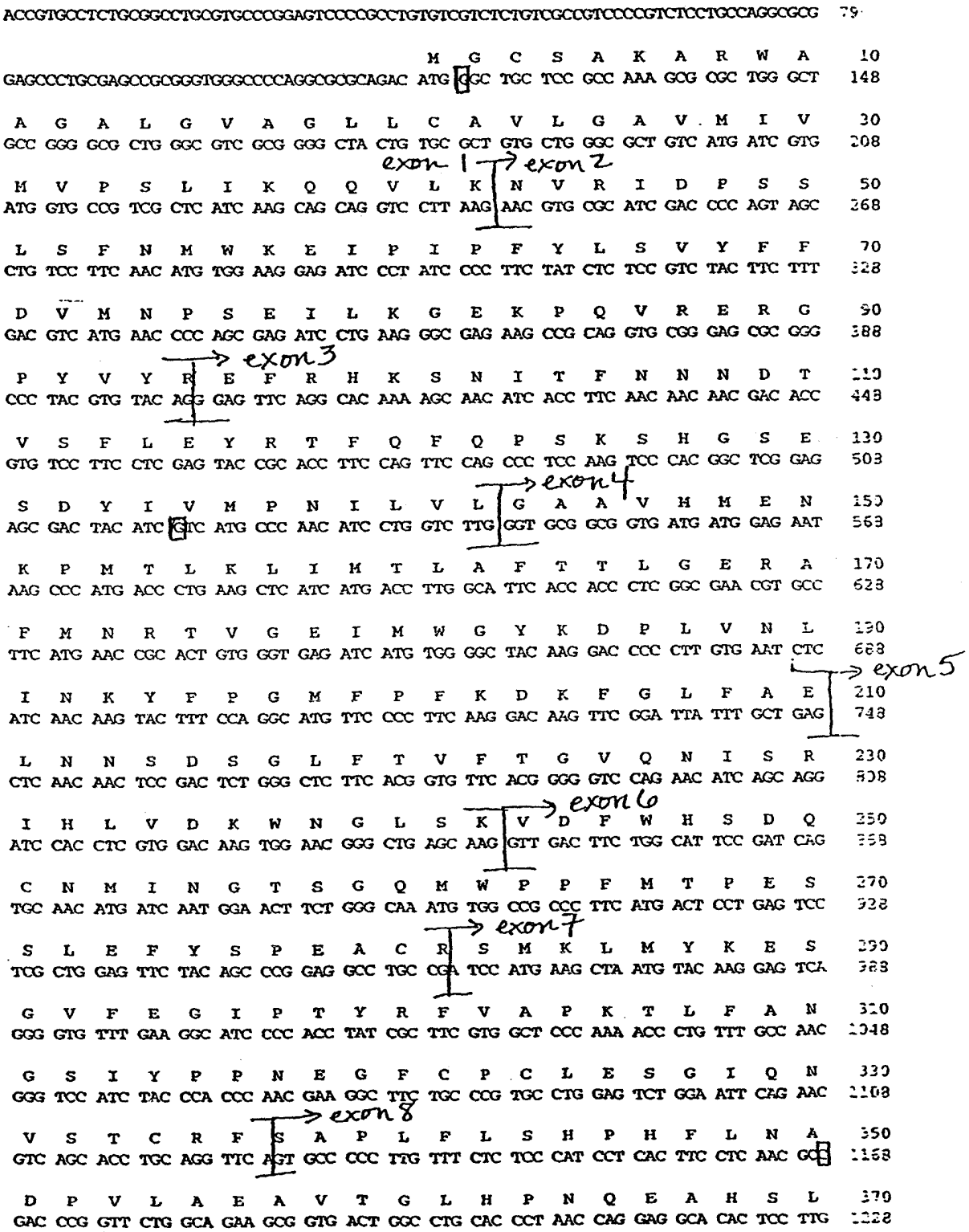
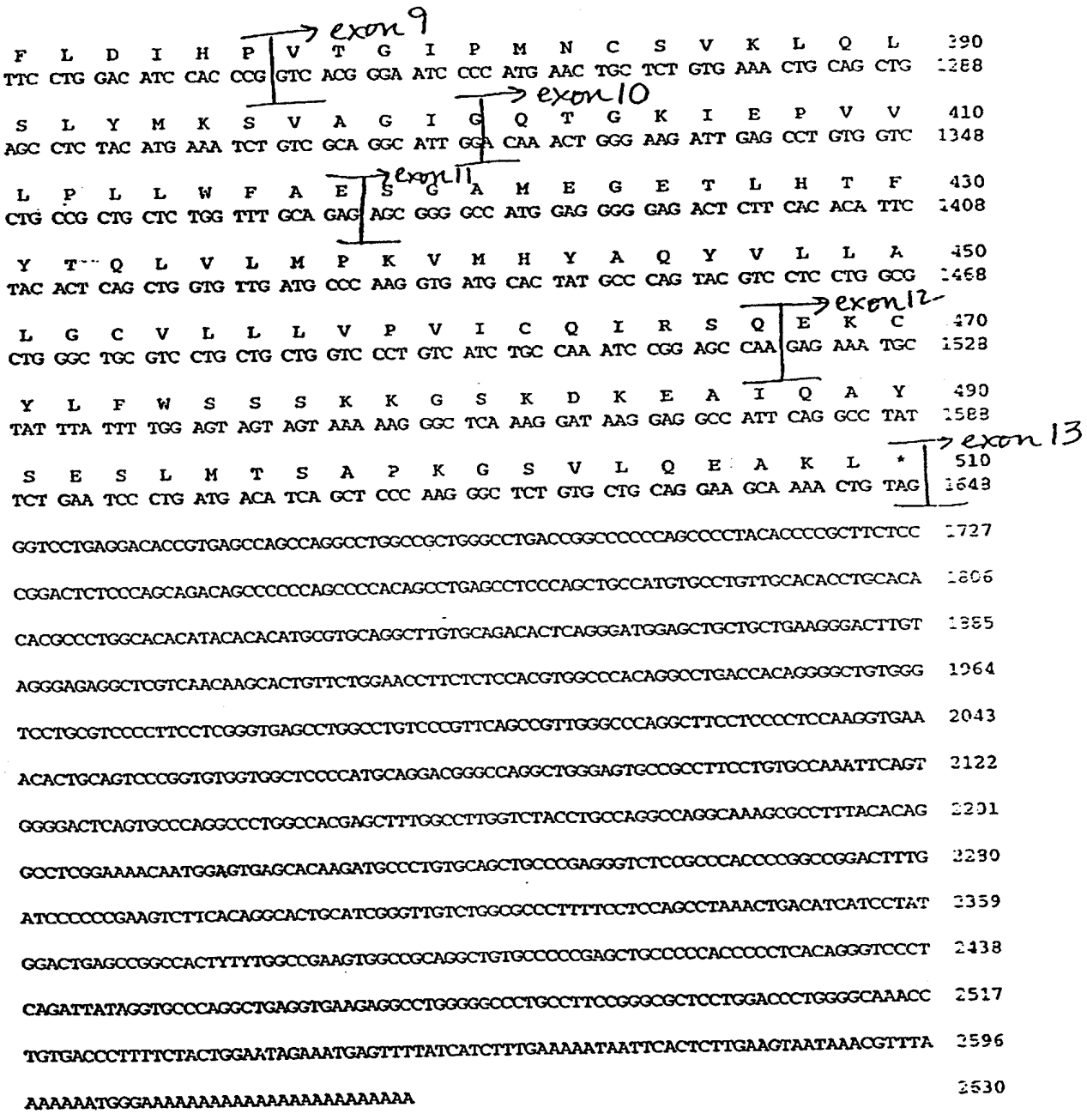


Figure 3B


exon 9
exon 10
exon 11
exon 12
exon 13
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S L Y M K S V A G I G Q T G K I E P V V 410
AGC CTC TAC ATG AAA TCT GTC GCA GGC ATT GGA CAA ACT GGG AAG ATT GAG CCT GTG GTC 1348
L P L L W F A E S G A M E G E T L H T F 430
CTG CCG CTG CTC TGG TTT GCA GAG AGC GGG GCC ATG GAG GGG GAG ACT CTT CAC ACA TTC 1408
Y T Q L V L M P K V M H Y A Q Y V L L A 450
TAC ACT CAG CTG GTG TTG ATG CCC AAG GTG ATG CAC TAT GCC CAG TAC GTC CTC CTG GCG 1468
L G C V L L L V P V I C Q I R S Q E K C 470
CTG GGC TGC GTC CTG CTG CTG GTC CCT GTC ATC TGC CAA ATC CGG AGC CAA GAG AAA TGC 1528
Y L F W S S S K K G S K D K E A I Q A Y 490
TAT TTA TTT TGG AGT AGT AGT AAA AAG GGC TCA AAG GAT AAG GAG GCC ATT CAG GCC TAT 1588
S E S L M T S A P K G S V L Q E A K L * 510
TCT GAA TCC CTG ATG ACA TCA GCT CCC AAG GGC TCT GTG CTG CAG GAA GCA AAA CTG TAG 1648
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CACGCCCTGGCACACATACACACATGCGTGCAGGCTTGTGCAGACACTCAGGGATGGAGCTGCTGCTGAAGGGACTTGT 1885
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TCCTGCGTCCCCTTCCTCGGGTGAGCCTGGCCTGTCCCGTTCAGCCGTTGGGCCCAGGCTTCCTCCCCTCCAAGGTGAA 2043
ACACTGCAGTCCCGGTGTGGTGGCTCCCCATGCAGGACGGGCCAGGCTGGGAGTGCCGCCTTCCTGTGCCAAATTCAGT 2122
GGGGACTCAGTGCCCAGGCCCTGGCCACGAGCTTTGGCCTTGGTCTACCTGCCAGGCCAGGCAAAGCGCCTTTACACAG 2201
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Figure 4

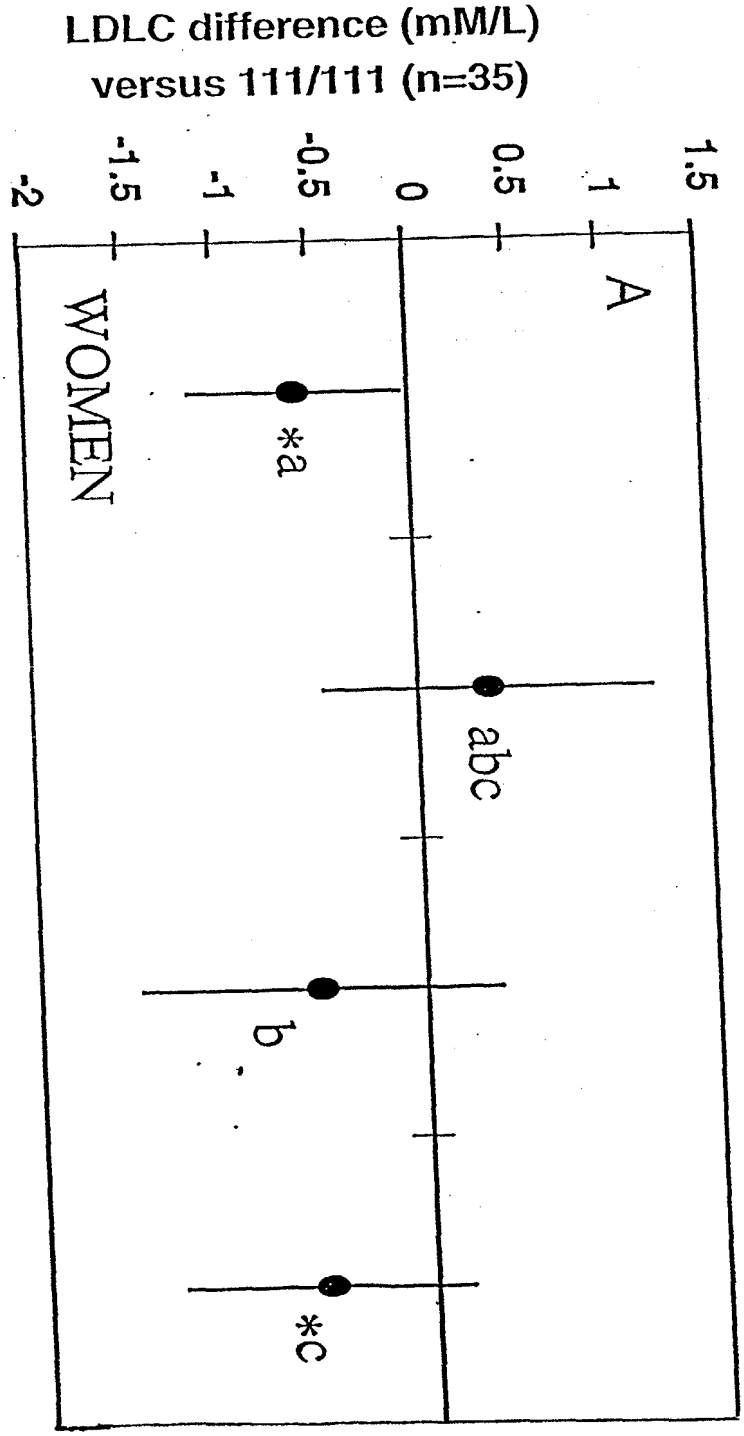

LDLC difference (mM/L)
versus 111/111 (n=35)
1.5
1
0.5
0
-0.5
-1
-1.5
-2
A
WOMEN
*a
abc
b
*c
112/111 (n=91)
121/111 (n=15)
211/111 (n=12)
112/112 (n=24)
Genotype

Figure 5

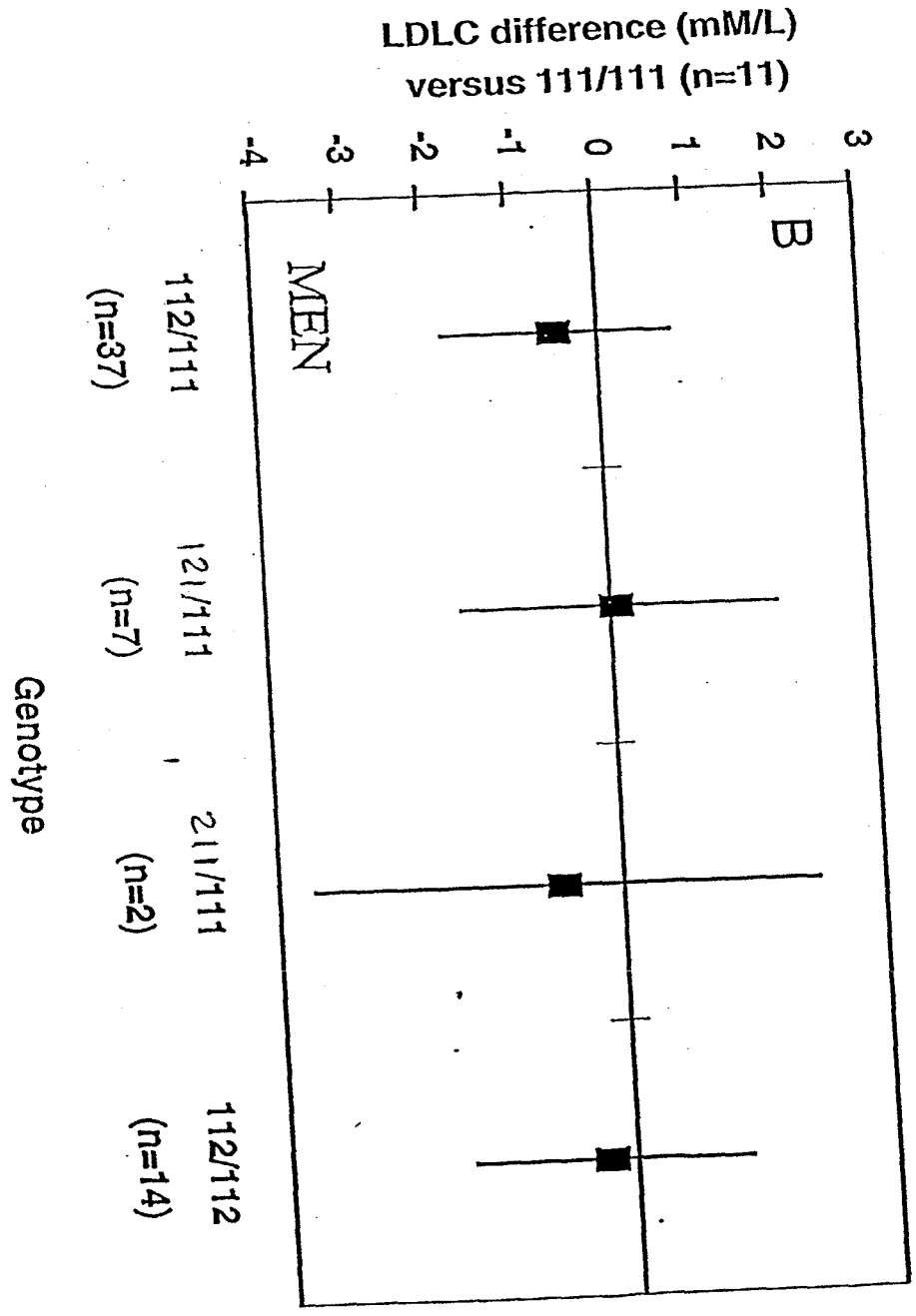

LDLC difference (mM/L)
versus 111/111 (n=11)
-4
-3
-2
-1
0
1
2
3
B
MEN
112/111
(n=37)
121/111
(n=7)
211/111
(n=2)
112/112
(n=14)
Genotype

Figure 6

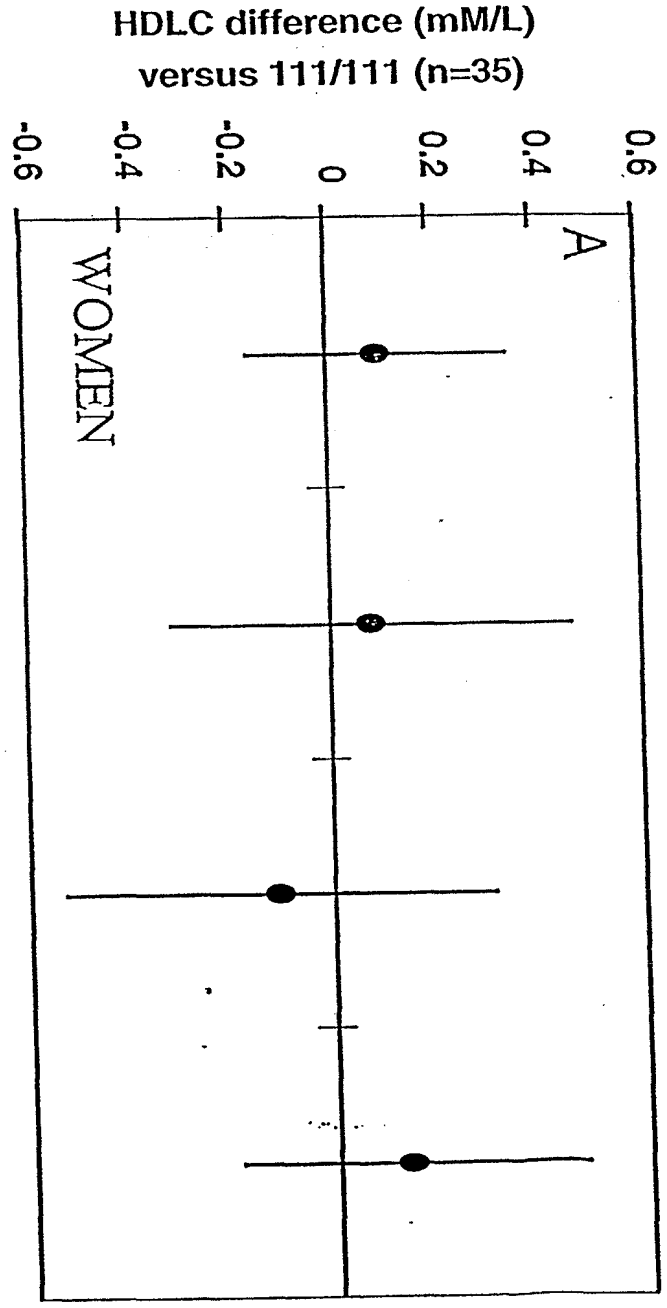

HDLC difference (mM/L)
versus 111/111 (n=35)
0.6
0.4
0.2
0
-0.2
-0.4
-0.6
A
WOMEN
112/111 (n=91)
121/111 (n=15)
211/111 (n=12)
112/112 (n=24)
Genotype

Figure 7

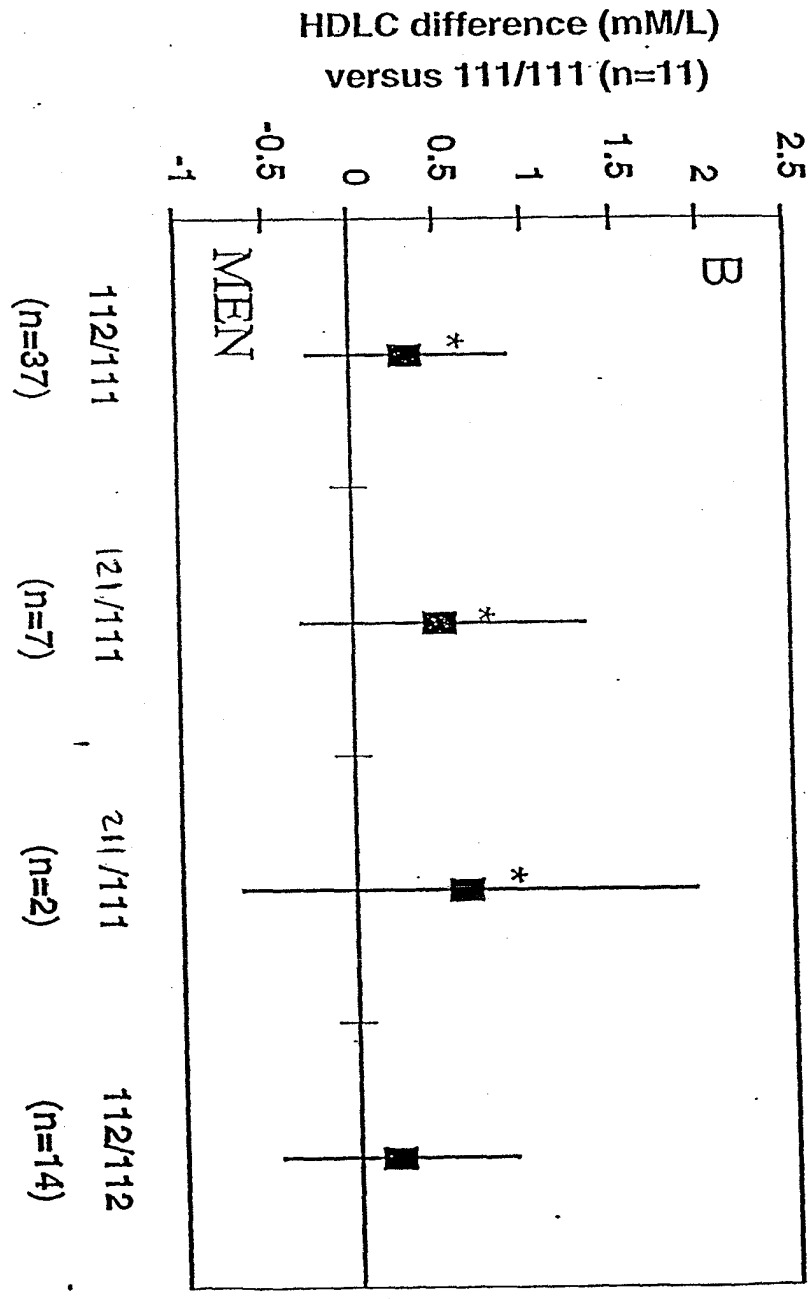

HDLC difference (mM/L)
versus 111/111 (n=11)
-1
-0.5
0
0.5
1
1.5
2
2.5
MEN
B
*
*
*
112/111 (n=37)
121/111 (n=7)
211/111 (n=2)
112/112 (n=14)
Genotype

Figure 8

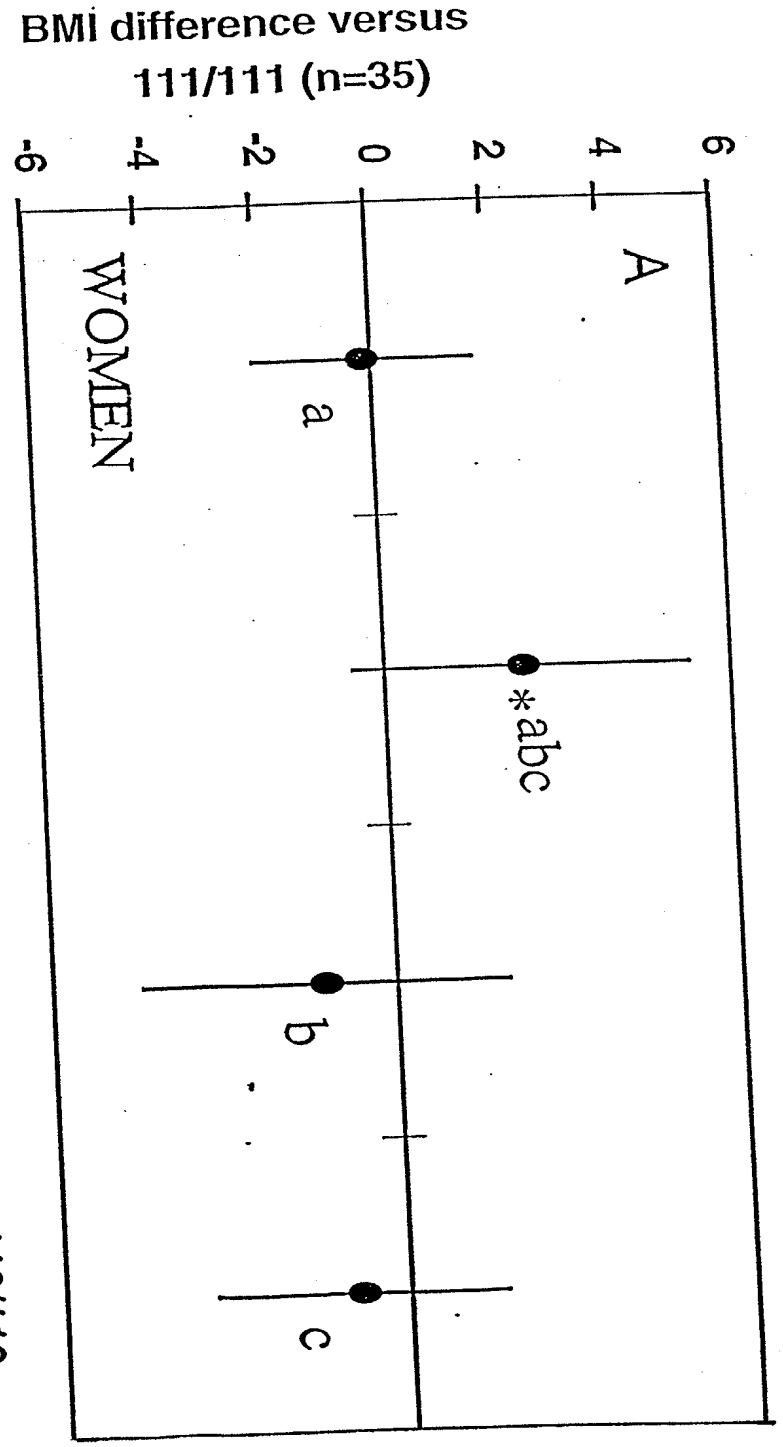
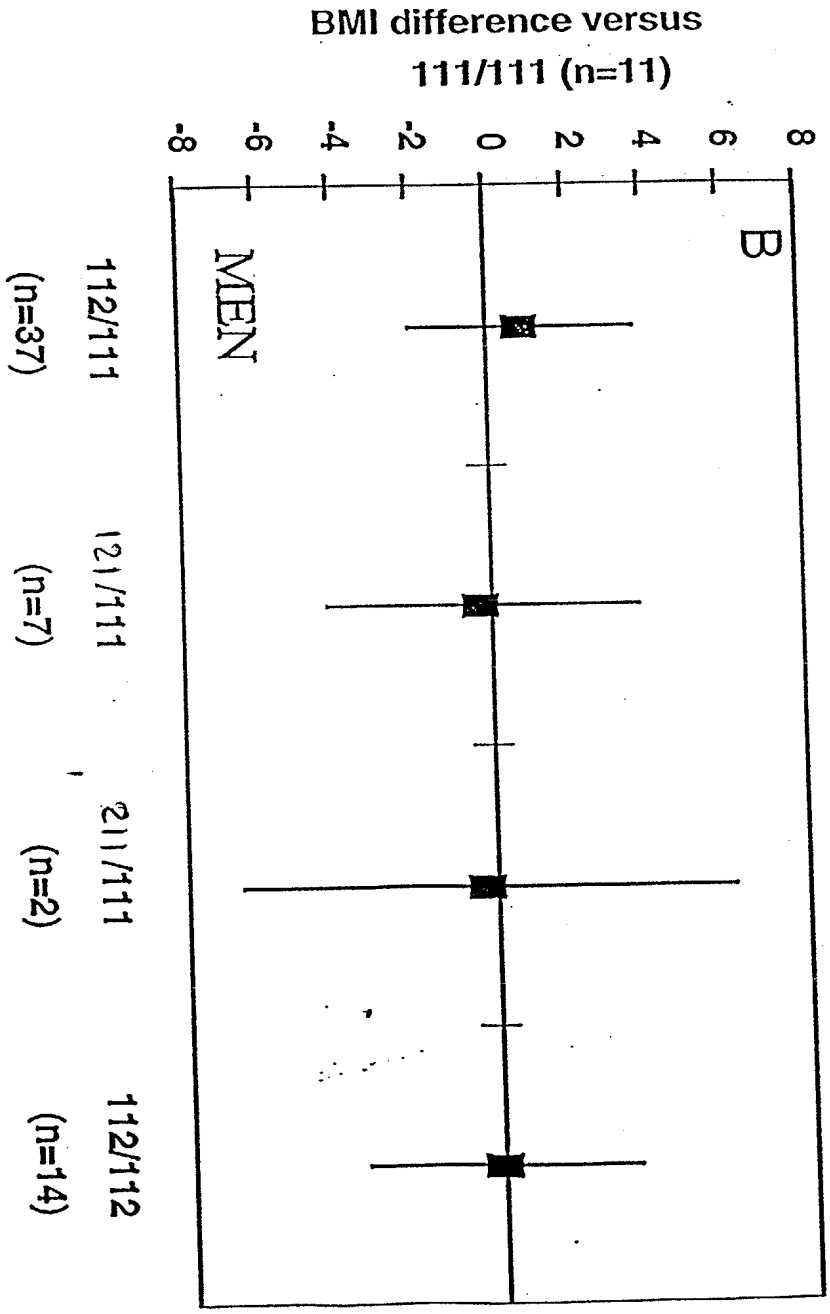

BMI difference versus
111/111 (n=35)
6
4
2
0
-2
-4
-6
A
WOMEN
a
*abc
b
c
112/111
(n=91)
121/111
(n=15)
211/111
(n=12)
112/112
(n=24)
Genotype

Figure 9


BMI difference versus
111/111 (n=11)
-8
-6
-4
-2
0
2
4
6
8
B
MEN
112/111
(n=37)
121/111
(n=7)
211/111
(n=2)
112/112
(n=14)
Genotype